



## TMPRSS6 gene

transmembrane protease, serine 6

### Normal Function

The *TMPRSS6* gene provides instructions for making a protein called matriptase-2. This protein is part of a signaling pathway that controls the levels of another protein called hepcidin, which is a key regulator of iron balance in the body. When blood iron levels are low, this signaling pathway reduces hepcidin production, allowing more iron from the diet to be absorbed through the intestines and transported out of storage sites (particularly in the liver and spleen) into the bloodstream. Iron is an essential component of hemoglobin, which is the molecule in red blood cells that carries oxygen.

### Health Conditions Related to Genetic Changes

#### iron-refractory iron deficiency anemia

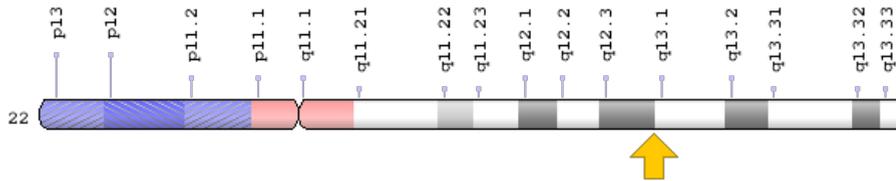
At least 40 mutations in the *TMPRSS6* gene have been found to cause an inherited form of anemia called iron-refractory iron deficiency anemia. This condition is characterized by a shortage (deficiency) of iron in the bloodstream that is resistant (refractory) to treatment with iron.

*TMPRSS6* gene mutations greatly reduce the amount of functional matriptase-2, preventing it from controlling hepcidin levels. The resulting elevation in hepcidin activity blocks the absorption of iron through the intestines and the release of iron from storage. When not enough iron is available in the bloodstream, less hemoglobin is produced and red blood cells cannot carry oxygen to the body's cells and tissues effectively. The shortage of oxygen causes the signs and symptoms of anemia, which can include tiredness (fatigue), weakness, and pale skin.

## Chromosomal Location

Cytogenetic Location: 22q12.3, which is the long (q) arm of chromosome 22 at position 12.3

Molecular Location: base pairs 37,065,436 to 37,109,701 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- matriptase 2
- matriptase-2
- membrane-bound mosaic serine proteinase matriptase-2
- transmembrane protease serine 6
- type II transmembrane serine protease 6

## Additional Information & Resources

### Educational Resources

- Biochemistry (fifth edition, 2002): Hemoglobin Transports Oxygen Efficiently by Binding Oxygen Cooperatively  
<https://www.ncbi.nlm.nih.gov/books/NBK22596/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TMPRSS6%5BTIAB%5D%29+OR+%28matriptase-2%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

### OMIM

- TRANSMEMBRANE PROTEASE, SERINE 6  
<http://omim.org/entry/609862>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_TMPRS6.html](http://atlasgeneticsoncology.org/Genes/GC_TMPRS6.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=TMPRS6%5Bgene%5D>
- HGNC Gene Family: Proteases, serine  
<http://www.genenames.org/cgi-bin/genefamilies/set/738>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=16517](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16517)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/164656>
- UniProt  
<http://www.uniprot.org/uniprot/Q8IU80>

## **Sources for This Summary**

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